Cancer genetics and its ‘different faces of autonomy’

DIRK STEMERDING* & ANNEMIEK NELIS†

*Department of Science, Technology, Health and Policy Studies, University of Twente, Enschede, The Netherlands
†Centre for Society & Genomics, Radboud University, Nijmegen, The Netherlands

ABSTRACT In this article we discuss the development of a practice of screening, preventive treatment, and presymptomatic testing for individuals at risk of Familial Adenomatous Polyposis (FAP), a specific hereditary predisposition for colon cancer. We describe this development as a process of co-evolution, showing how this practice has been gradually taking shape in a new network of actors, routines, rules, institutions and technologies. We further argue that, looking at the emergence and transformation of this practice, we can distinguish two different regimes: a regime of prevention and a regime of self-determination. In each of these regimes the autonomy of patients and individuals at risk is shaped in a different way, that is, through a different complex of ideals, procedures, institutions, technologies, and routines. In our view, the interference between these two regimes is an important characteristic of the emergent new genetics and is reflected in the growing debate about non-directivity in genetic counselling. However, as our argument implies, when facing the challenges of the new genetics we should not restrict the debate to the quality and ethics of counselling, but extend our view to the whole complex of elements and activities which shapes individual autonomy in the context of different regimes.

Introduction

In today’s health care practices, and the writing about these practices, patient autonomy and self-determination is held in high esteem. A good doctor leaves patients room for choice. It is a doctor’s duty to respect the autonomy of his or her patients and to weigh this duty against other moral duties such as the duty to do no harm and to benevolence. The increasing attention for patient choice and the right to self-determination often is described in terms of a changing morality: in modern society we value individual choice and a liberal mode of decision making (Beauchamp & Childress, 2001).

The emphasis on choice and decision-making is particularly manifest in the field of reproductive choices. The development of new technologies such as prenatal testing, IVF and DNA testing has played an important role in the articulation of the individual responsibility for reproductive choices. In clinical genetics
this has resulted in a medical practice in which patients are addressed primarily as individuals who have to make informed choices and in which non-directiveness is the norm (Bosk, 1992; Steendam, 1996; Clark, 1998). In centres for clinical genetics, genetic diagnosis is embedded in a practice of counselling in which facts deemed relevant are separated out as ‘information’, that is handed to clients along with a few courses of action formulated as possible alternatives and between which clients are requested to choose. The establishment of clinical genetics thus created a particular space for individual decision-making in which, from the 1990s onwards, new options for genetic testing on the basis of DNA-diagnosis have been introduced and evaluated primarily as bringing along new opportunities of choice (Nelis, 1998).

However, in the context of emergent practices of ‘new genetics’ in which individuals are not only presented with more and more information about future health risks, but also with related options for prevention, we see a growing debate about the merits and limits of a non-directive approach (Biesecker, 1998; de Wert, 1999; Weil, 2003; Resnik, 2003). Depending on the risks involved, and the availability and burden of preventive options, unsolicited recommendations about monitoring, medical interventions or lifestyle are generally deemed more justified. From this point of view, future practices of genetic testing thus will (have to) be shaped by more pro-active and directive approaches. A characteristic feature of this debate about non-directiveness in the era of the new genetics, is a strong focus on the practice of genetic counselling and the professional ethos of the counsellor. The normative debate is dominated by a behavioral perspective focusing on the question how to respect individual autonomy and to create, in the context of genetic counselling, optimal circumstances for decision-making about genetic testing, medical surveillance, (prophylactic) treatment, and long-term changes in risk-related health behaviors (Grosfeld et al., 2000; Lerman & Croyle, 1996).

In this article we want to consider the issues of autonomy and non-directiveness from a broader perspective, in which we relate the morality of (non-)directiveness not only to the duties of counselling, but to the whole network of technologies, institutions, rules, routines, human actors, interactions and interventions constituting a medical practice. The emergence of the new genetics we see as a process of change in which new technological options and various other elements and activities constituting medical practices shape each other in new ways. As we have discussed elsewhere, this process may be described in terms of co-evolution (Stemerding & Nelis, 2004, see also Stemerding et al., 1997; Bourret et al., 1998; Bourret, 2005). From this co-evolutionary perspective, we will focus in this article on the various ways in which the autonomy of patients and individuals at risk is embodied in and shaped by new emerging practices of screening, prevention and predictive testing in the field of cancer genetics.

The analysis in this article is based on a study of the way in which in the Netherlands a practice has emerged of screening, preventive treatment, and presymptomatic testing for individuals at risk of Familial Adenomatous Polyposis (FAP), a specific hereditary predisposition for colon cancer. Our study consisted
of an extended literature review, including the various publications about FAP which started to appear in the Dutch Journal of Medicine from the beginning of the 1980s, and the most relevant publications from the international literature. In addition we interviewed (key) actors who were actively involved in the field, as researcher, medical specialist, social worker, or clinical geneticist. The interviews have been conducted in the period 1996–1997.

In the following, we describe three different periods that mark the development of a practice of screening, prevention and treatment of FAP. First, we describe how in the Netherlands, in the 1980s, a preventive practice was created of screening and treatment which involved both patients and family members at risk. Next, we describe how this practice was organized at a more collective level through the establishment of a national registry of FAP families. Finally, we describe how this practice was further extended with the development and introduction of presymptomatic DNA-diagnostic tests in the early 1990s. In the second half of this article, we will analyse the development of the extending FAP-network in terms of two co-existing regimes: a regime of prevention and a regime of self-determination. In each of these regimes the autonomy of patients and individuals at risk is shaped in a different way, that is, through a different complex of ideals, procedures, institutions, technologies, and routines.

FAP in the early 1980s: the emergence of a screening practice

In 1981 a collection of three articles and a commentary appeared in one of the issues of the Dutch Journal of Medicine, focusing on the clinical experiences with a rare, dominantly hereditary disease, called polyposis coli or familiar adenomatous polyposis (FAP). In each of the three articles an elaborate description was given of the history of the disease in a particular family (de Ruiter & den Hartog Jager, 1981; Vissers & Stroosma, 1981; Raymakers et al., 1981). One of the articles opened with the story of a 27-year old woman who consulted the clinic because a 34-year-old cousin of hers had been recently identified as a FAP patient after the diagnosis of colon cancer (de Ruiter & den Hartog Jager, 1981). The mother of the woman had died from colon cancer when she was 43, a few years after the colon partly had been removed because of a malignant polyposis. The authors of the article observed with regret that clinical examinations of relatives had not been undertaken at the time, although it was known that a grandmother and a great-grandfather also had died from ‘cancer of the colon’. The woman who was consulting the clinic had no symptoms, but inspection of the colon revealed many polyps resulting in a subsequent diagnosis of FAP and in the decision to completely remove the colon. The article then continued with the case of a younger sister of the woman, who similarly displayed no symptoms, but who also had her colon removed after investigation had indicated polyposis. Examination of other brothers and sisters only revealed a few polyps in the colon. In these cases, as the authors point out, the examination had to be repeated every year. In conclusion, the article reports the results of examinations that had
been carried out in 39 people, spanning two generations of the family. Polyposis was found in 12 cases, and colon cancer had already developed in four of these. Four members of the family refused the invitation to undergo examination.

The collective publication of the three articles and the accounts given by the authors of the diagnosis, treatment and screening in families where FAP is found may be seen as an event which marks the emergence of a specific clinical practice. That is, a practice in which the professional responsibility of the medical specialist—an internist or gastro-enterologist—cannot be restricted to the individual patient, but should extend also to the health and survival of the patient’s relatives (see Fig. 1). When a patient consults the clinic with specific symptoms and the diagnosis of FAP is made, in most cases a fatal colon cancer will have already appeared. Thus, as the authors of the articles point out, it is of vital importance to trace the families in which FAP is found and to screen the members of these families every two or three years, beginning from about the age of 10. As soon as in the colon more than one hundred polyps are found, the diagnosis of FAP should be made, and it is only by complete removal of the colon that the development of cancer can be prevented. Even then, regular screening will remain necessary. If, on the other hand, members of the family are still free of symptoms between the ages of 40 and 50, the appearance of polyposis can be reasonably excluded and screening may be terminated.

The conclusions and recommendations of the authors clearly were intended to promote a standard that, at the beginning of the 1980s, often was more theory than practice. At that time, it was clearly considered against due practice to refrain from an extensive family anamnesis when the diagnosis of FAP had been made. However, in everyday clinical practice this kind of family anamnesis did not always take place, which is explained, in one of the articles, by a general lack of knowledge of the serious consequences of the disease (Vissers & Stroosma, 1981). Even when an extensive mapping of the family history of the patient followed the diagnosis of FAP, it was often considered to be an impossible task for the individual specialist to actively approach all the family members involved. And, as far as family members were actually approached, the specialist could not

![Figure 1. Emergent FAP-network at the end of the 1970s.](image-url)
always be sure that they would return regularly for periodical screening. Thus, in a commentary on the three articles mentioned above, published in the same issue of the *Dutch Journal of Medicine*, it was observed that:

Detective-like genealogical investigations, the psychological burden experienced by people who feel completely healthy and yet face the prospect of invasive examinations of the colon, and the not always interesting task to screen a fairly large number of people who have no symptoms, require a great and unremitting enthusiasm and dedication of those who undertake to follow a family with a history of polyposis. (van Slooten, 1981, p. 1763)

Hence, the author of the commentary argued for the establishment of a centralized national registry, which would send out reminders to medical specialists each time a person at risk had to be called up for screening. The results of the screening were to be returned to the registry. If no results followed, the organization should be enabled to take further action in order to safeguard the care for those at risk and to obtain certainty about their condition. With his plea, the author actually repeated a message that he had voiced already in the same journal more than 25 years earlier (van Slooten, 1953). This time, however, the argument would find an audience.5

Late 1980s: the establishment of a national registry

At the end of the 1980s, a patient who consulted the clinic with symptoms of FAP would encounter a practice that indeed was different from what we have seen before. Now, the medical specialist in attendance of the patient not only had to inform the patient about the hereditary nature of the disease and about the importance of screening family members, but also could refer the patient to the national *Foundation for the Detection of Hereditary Tumours* (STOET). In 1983, this foundation was established by a number of specialists involved in the treatment of patients and their families suffering from hereditary cancers (one of the founders was the author of the commentary quoted above). In 1985 the Foundation started a national registry of families with a history of FAP, thus aiming to promote screening in high-risk families, to guarantee the continuity of screening, to collect data for scientific purposes, and to offer advice about diagnosis, treatment, methods of screening, and genetic services for counselling (Vasen *et al*., 1988).

The result of this development was a more extended practice of diagnosis, treatment and screening of FAP, in which every patient is reported by medical specialists to the Foundation for the Detection of Hereditary Tumours (see Fig. 2). A social worker of the Foundation approaches the patient and with his or her help draws up a family tree which makes it possible to trace the history of the disease and to identify members of the family who are at risk. The patient is asked to inform relatives at risk and to urge them to have themselves screened. If they agree, family members are approached by the Foundation...
with a request for registration. In this way, nearly all FAP families in the Netherlands have been registered, amounting to a few hundred families (Annual Report STOET, 1994). Personal and medical information is collected from those who have registered and, through a system of reminders, specialists are notified when individuals should be called up for screening. If no screening results are reported back and upon inquiry it appears that someone did not turn up for screening, the registry will send out a request to the family doctor to take action and to remind this person that screening is of vital importance.

While the care for individuals at risk of FAP initially strongly depended on the enthusiasm and efforts of individual specialists and on the awareness of those at risk, it is now the Foundation for the Detection of Hereditary Tumours which has assumed the responsibility for the organization and continuity of screening and which ‘will put all efforts in encouraging [registered] individuals to comply [to regular screening]’ (Annual Report STOET, 1994, p. 5). Indeed, as those working for the Foundation point out, in order to motivate family members to participate in a screening programme, good information and a personal approach including visits at home are necessary:

Visiting people at home works out very well. You have to adapt yourself to their environment and there is more time for explaining things. It is also more easy to persuade people of the benefit of screening. (An interview with a social worker of the Foundation)

The finding of many at-risk family members who had not previously been screened is considered as a clear proof of the value of a national registry (Vasen et al., 1990).
Moreover, through the establishment of a registry, a lot of information can be collected which not only facilitates the organization of a screening programme, but which also creates possibilities for a systematic follow-up and an evaluation of its results. For that purpose, two national working groups on FAP, involving various forms of expertise, collaborate with the Foundation in the organization of studies and the establishment of guidelines (Vasen et al., 1988). Thus, through the efforts of the Foundation, local practices of early detection and prevention have become part of a larger network in which these practices are organized and regulated on a national scale.

**Early 1990s: the advent of DNA-diagnosis**

In the early 1990s, a patient who consulted the clinic with symptoms of FAP again would encounter a practice that had been extended with new elements—new technologies, rules and organizations. Through its well-kept database the Foundation for the Detection of Hereditary Tumours had become a major resource for research on FAP, and in particular research on genetic markers as was done at the genetics laboratory of the University of Leiden. Indeed, according to the clinicians involved:

> Strong relationships to the gastroenterological clinic and to the Foundation for the Detection of Hereditary Tumours (STOET), on the one hand, as well as the creation of a strong colorectal cancer genetics group of basic scientists, on the other, laid the basis of (genetic) studies on FAP over the past 15 years. (Griffioen et al., 1999, p. 127)

In 1989 researchers in Leiden succeeded in finding genetic markers on both sides of the so-called APC (adenomatous polyposis coli) gene (Tops et al., 1989). This finding was soon followed by the identification of the APC gene and opened the possibility of presymptomatic DNA-diagnosis, whereby members of a FAP family may be informed about their individual risk-status (Vasen & Müller, 1991). However, in the Netherlands, DNA-diagnosis is made available only through a network of regional clinical genetics centres. Thus, with the advent of DNA diagnosis of FAP, genetics laboratories and clinical genetic centres became part of the network in which the practice of diagnosis, treatment and screening of FAP took shape (see Fig. 3).

A new patient now not only will be reported by the medical specialist to the Foundation for the Detection of Hereditary Tumours, but also will be referred to a clinical genetics centre for DNA-analysis. Again, a counsellor of the clinical genetic centre will draw a family pedigree in order to identify relatives who may be at risk of developing the disease. Members of the family then have the opportunity (after having been informed by the patient) to be referred to a clinical genetics centre, which may offer them presymptomatic DNA-diagnosis. Those who accept the offer and are diagnosed as carriers, have the certainty that they will get FAP and that regular screening is the only way to escape from an early and deadly cancer. Of those diagnosed as carriers most, if not all, will have themselves
registered in the national registry of the Foundation for the Detection of Hereditary Tumours. Those however who are diagnosed as non-carriers are excluded from risk and thus may abstain from participating in a burdensome and protracted screening programme. In other words, DNA-diagnosis opened the possibility to divide a known population at risk of developing FAP into a carrier group which can be followed with traditional clinical screening methods, and a non-carrier group which may be excluded from risk and relieved from participating in a screening programme. For those appearing to be carriers, DNA-diagnosis may have additional value in decisions about prophylactic interventions, and is available in the form of prenatal diagnosis.\(^6\) Thus, options for DNA-diagnosis have been readily incorporated in clinical practice as a diagnostic tool which contributes to more efficient and improved forms of preventive care (Menko et al., 1999a, 1999b; van der Luijt et al., 2000; King et al., 2000).

**A network of co-existing regimes**

In the previous sections we have described the emergence of a national screening programme in the field of cancer genetics as a process of co-evolution, showing changing configurations of actors, routines, rules, institutions and technologies in which a particular case of ‘new genetics’ has been gradually taking shape. We have depicted these changing configurations as an extending network, which involved new actors—family members potentially at risk, social workers, a national registry, clinical geneticists—and which established new alignments between these actors. In this network, the provision of information to family

---

*Figure 3. Extended FAP-network in the 1990s.*
members about the hereditary nature of the disease, the establishment of family
trees, the collection of medical data, the offer of DNA-diagnosis and clinical
screening, became standard elements of the activities of the medical specialist,
social worker or clinical geneticist. The data collected through this (net)work,
by the Foundation for the Detection of Hereditary Tumours, facilitated not
only the organization of a national screening programme, but also the systematic
monitoring of its effects and the development of guidelines to be observed in
practices of diagnosis and screening. Thus, the extending FAP-network embodied
a long envisaged task to improve the management of polyposis as a hereditary
disease.

Our previous account of the emergence of a FAP-network not only describes
the mutual shaping or co-evolution in which FAP was transformed from a fatal
disease of the colon into a hereditary disorder with preventable consequences
(Palladino, 2002). It also shows how this process of co-evolution crystallized
into a specific pattern of roles and responsibilities embodied in the emergent
configuration of the network. In the early 1980s, as the publications in the
Dutch Journal of Medicine show, medical specialists already were expected to
inform FAP-patients about the hereditary nature of the disease, and the conse-
quent implications for members of the family. The responsibility of the physician
should not stop with the treatment of a patient, but should also extend to the
patient’s family. Relatives had to be informed about potential risks and if
necessary, according to the ruling standards of the time, considered for regular
screening. With the establishment of a national registry, the responsibility of
individual specialists to offer information and care to the family of patients
became institutionalised on a more collective level in the working practices,
data-base and protocols of the Foundation for the Detection of Hereditary
Tumours. In other words, through the efforts of the Foundation, practices of
eye detection and prevention no longer depended primarily on local initiatives,
but had become part of what we might call a new cancer genetic regime of
prevention.7

When, at the end of the 1980s, researchers in Leiden found genetic markers
on both sides of the so-called APC gene, followed by the identification of the
gene in the early 1990s, it became possible to identify gene carriers in at-risk
families. However, the actual provision of a DNA-test did not come within the
province of the gastro-enterologist or the Foundation for the Detection of
Hereditary Tumours. It was the Leiden centre for clinical genetics that organized
and facilitated the introduction of DNA-diagnosis in FAP-families. In the
Netherlands, from the 1970s, specialized centres for clinical genetics have
been assigned a privileged position in offering genetic counselling and genetic
testing, and function as ‘gatekeepers’ for those seeking genetic consultation
and diagnosis (Nelis, 2000). In these centres, genetic diagnosis is embedded
in a practice of counselling in which autonomous decision-making of patients
and individuals at risk is the guiding principle. The responsibility for decisions
and actions to be taken is delegated primarily to the individual asking for infor-
mation and advice. Thus, when providing information, counsellors consider it as
their task to be neutral and non-directive.\textsuperscript{8} In the practice of genetic counselling the principle of informed decision-making is also upheld by organizational and material provisions, like the relatively long time available for each consult, the obligatory time-frames between consults when clients have to make important decisions, and the extensive documentation of consultations that counsellors provide to their clients.

In other words, when DNA-diagnosis became available for those at risk of developing FAP, it was embedded in a practice that was coordinated through an infrastructure of independent clinical genetic centres and that already constituted a regime of its own. In this regime, self-determination was the guiding principle that defined the roles and responsibilities of the actors involved.\textsuperscript{9} In this context it is interesting to see how medical specialists, in the early 1990s, considered the prospects of DNA-diagnosis in the field of cancer genetics. They emphasized, first of all, the promise of improved forms of preventive care, but also pointed out that:

> With the new diagnostic options, decision-making will become more and more difficult for members of the family, and the ethical aspects of genetic counselling will become of paramount importance. The individual at risk should have complete freedom to decide whether the advantages of testing outweigh the disadvantages. Non-directiveness has to be taken here as a basic principle. (Vasen & Müller, 1991, p. 1622)

This comment clearly reflects the history and position of clinical genetics as a practice in which DNA-diagnosis is made available to patients and individuals at risk primarily as an opportunity of (informed) choice. In other words, in our account of the emerging FAP-network, the introduction of options for DNA-diagnosis not only involved the extension of this network with a few new elements, but also created a situation of co-existence in this network of two different regimes. Indeed, as we will see below, from the 1990s onwards, FAP patients and individuals at risk were supposed to follow a trajectory in which they were addressed both as subjects who need preventive care and as subjects who have to deal with (new) opportunities of choice.

***Different faces of autonomy***

The point we want to focus on in our analysis of the emerging FAP-network is how changing configurations of actors, routines, rules, institutions, technologies and responsibilities are related to changing and conflicting notions of autonomy of patients and individuals at risk. Our description of two co-existing regimes embodied in the FAP-network clearly relates to the debate about non-directiveness that has been raised in the context of the new genetics. In this debate, Biesecker and others have argued that once there is sufficient evidence that a disease can be effectively treated or prevented, decisions about genetic tests will come to be regarded as medical, rather than as personal decisions, and medical providers will likely be more directive in their approach (Biesecker, 1998; see also de Wert, 1999).
In a similar vein, Lerman and Croyle, have questioned the appropriateness of the traditional non-directive stance, arguing that in cases such as breast and colon cancer, for which effective surveillance methods are available, the absence of encouragement and recommendations by providers may be morally unjustifiable (Lerman & Croyle, 1996). The visions of these authors are clearly reflected in the practices that we have described as a regime of prevention, putting all efforts in encouraging individuals to comply, in contrast to a regime of self-determination which embodies the non-directive stance.

Our notion of regime however explicitly refers to a whole configuration of actors, routines, rules, institutions, technologies and responsibilities constituting a medical practice, whereas the debate about non-directiveness almost exclusively focuses on the practice of counselling and the professional ethos of the counsellor. In her reflections on future directions in genetic counselling, Biesecker (1998), for example, has observed that genetic counsellors may come to consider families, rather than individuals, as their clients. A nice illustration of this point is the FAP-network, in which the professional responsibility of the medical specialist actually has been extended from the individual patient to the patient’s relatives. However, from our point of view, this responsibility does not only involve a particular ethos of counsellors. It also has been shaped and institutionalized at a more collective level of a regime, through organized practices of information, family investigation, registration, screening and data-collection.

Likewise, we may consider the issues of autonomy and non-directiveness from this more inclusive point of view, as Schermer (2001) has done in an empirical study of decision-making practices in a hospital setting. In her study Schermer shows that medical decision-making in daily care-settings is an ongoing and diffuse process. Decisions are rarely made at one time, in one location or by one person, let alone by the individual patient. On the contrary, ‘there were many moments, with many smaller and bigger decisions clustering together’ (Schermer, 2001, p. 79) that jointly produced a particular trajectory or action. Schermer thus includes in her analysis the whole setting in which decision-making takes place, rather than focusing on isolated instances of physician-patient interaction. From this point of view, patient autonomy does not so much appear as a singular act of self-determination, but as being shaped by the various activities, rules, routines and procedures which make up everyday medical practice. Autonomy, as Schermer puts it, may have ‘different faces’.

In our account of the emerging FAP-network we have also focussed on the broader setting in which decision-making is part of a trajectory of various activities involving, at different moments and in different locations, both physicians, social workers, counsellors, patients and individuals at risk. In this way we have traced the changing configurations of the FAP-network, and we have distinguished in this network two different, co-existing regimes. By following the trajectory of actors through the configurations of these different regimes, we now can see how, in these regimes, the autonomy of patients and individuals at risk is ‘configured’ in different ways as well.
Autonomy in a regime of prevention

Schermer’s description of the practices and routines of hospital care obviously has much in common with our previous account of the ways in which patients and individuals at risk move through a FAP-network constituting, what we have called, a regime of prevention. As Schermer (2001, p. 82) observes, in the daily care-setting of a hospital, ‘considerations concerning a patient’s well-being’ seemed to be ‘far more important than considerations concerning patient autonomy’. As we have seen, the cancer genetic regime of prevention that is embodied in the FAP-network is shaped by strong links between clinical care, screening, registration and monitoring of both patients and their family members, with the aim to improve the chances of a healthy life for everybody who is at risk. Thus, in accordance with Schermer’s observation, we find in this regime of prevention a strong focus on the well-being of individuals at risk. Indeed, the primary aim of the Foundation for the Detection of Hereditary Tumours is to guarantee optimal care and so it does everything in its power to encourage individuals to cooperate. In this context the notion of individual autonomy primarily appears as a boundary that one should respect. As one of the social workers of the Foundation explains:

Because of privacy regulations we are not allowed to approach members of the family without their personal consent. Thus patients are invited by the Foundation to inform relatives at risk. Personal contact with patients at home makes it easier to persuade them that it is necessary to inform other members of the family and also to convince them of the benefit of screening. Sometimes, when patients are reluctant, the family doctor is called in. If members of the family don’t want to be informed, then there is nothing more to be done. (An interview with a social worker of the Foundation)

Although professionals in the FAP-network thus occasionally refer to notions of autonomy and choice, the organization of screening and medical decision-making in a regime of prevention is governed by judgements, routines and standards in which the medical well-being of patients is clearly assigned priority over other considerations.

In her study of decision-making practices, Schermer also observes that when the preferences of patients are taken into account, medical professionals often refer to what they presume patients in general deem as important, that is, to the image of the average or ‘standard’ patient (Schermer, 2001, p. 99). This observation likewise applies to the efforts of those involved in the FAP-network. As medical specialists have been increasingly convinced of the necessity to regularly screen members of families at risk, they have created new courses of action for patients and their relatives, and thus also new opportunities of choice. However, in adhering to the rules and standards of a regime of prevention, medical specialists are not seeking more room for choice, but are seeking opportunities to improve care. The course of action indicated is defined by the image of
a standard patient who needs, first of all, preventive care. Indeed, diagnosis, treatment and screening are seen as matters of (early) death or (longer) life and decisions about preventive measures are primarily perceived as medical issues. Although in this context, autonomy and choice are still considered valuable and indispensible notions, decision-making will be generally discussed and presented in terms of necessary interventions about which there is little scope for choice. Thus, reflecting on his discussions with FAP-patients in the consultation room, a gastro-enterologist comments:

The days that the doctor knew best are long gone. Choices and risks related to the timing and nature of surgical interventions have all to be clearly discussed with the patient. There are a lot of things that have to be considered, including of course the wishes of the patient. . . . Of course, when a colon is full of polyps, it is our task to deliver the message, to say what must be done, and that is, you know, what always will be done. (An interview with a gastro-enterologist at a university hospital)

Of course, not everybody needs to conform to the roles and responsibilities of the standard patient. Sometimes a patient is not prepared to inform other members of the family. And not everyone at risk really wants to be informed, or will turn up for screening. In living their lives and their disease, people thus may choose to follow different trajectories. However, with the emergence of a practice of clinical screening and its development into a cancer genetic regime, some trajectories have been made more comfortable and predictable than others. In the early 1980s it required a great deal of effort for a medical specialist to maintain a programme of screening that would allow every individual at risk to be informed and undergo regular examination. Today, a national registry, social workers who visit patients at home, information leaflets, a system of reminders, working groups, survival rates, guidelines, form the constitutive elements of a regime of prevention, offering patients and relatives at risk a course of action that is difficult for them to refuse.

Autonomy in a regime of self-determination

In addition to the clinic and the registry, clinical genetics centres have become, from the beginning of the 1990s, part of the emerging FAP-network. From that time on a patient or individual at risk may be referred by a gastro-enterologist or surgeon to a clinical genetics centre, and then will move along a trajectory structured by a regime of self-determination. In this regime too, the roles and responsibilities that are ascribed to patients and individuals at risk are shaped by standards and routines indicating the best course of action in the process of medical decision-making. Through a practice of counselling, supported by various procedural, organizational and material provisions, the responsibility for decisions and actions to be taken is delegated primarily to the individual coming for a test. Thus, when an individual at risk of FAP consults a clinical
According to the protocol, applicants for a pre-symptomatic test first see a clinical geneticist and a psychologist. The clinical geneticist discusses all the relevant aspects of the disease and also examines, of course, the personal reasons why someone is coming for a pre-symptomatic test. Then there is a meeting with one of the psychologists who discusses the various implications of pre-symptomatic testing. This is followed by a four-week period to think the matter over, after which the applicant returns for a final discussion about the test. When all questions have been answered and someone has come to a decision, blood will be taken. Then again there is a meeting with the psychologist in order to prepare someone for the test result. The test takes about three months. When the outcome is available, everything of course will be discussed again. . . . (An interview with a clinical geneticist at a regional clinical genetics centre)

In a regime of self-determination we find another image of the standard patient as someone who has a great stake in self-determination and non-interference in dealing with (new) opportunities of choice. Thus, in the day-to-day practice of clinical genetics, everything is done to uphold non-directiveness and informed freedom of choice. But, as we have noted above, the presumptions made about the values and preferences of the standard patient need not always match with the wishes or views of the individuals involved. For example, in moving through the FAP-network, many patients or individuals at risk see the clinical genetics centre as ‘just a stop on their way to the surgeon’ (Dudok de Wit, 1997, p. 179). Being referred by their gastro-enterologist or surgeon to know if screening is (still) necessary, they experience their choice as a purely medical decision, and sometimes feel annoyed by appointments with a clinical geneticist or psychologist taking time to explain the implications of predictive testing.

These experiences in the FAP-network clearly illustrate a point that has stirred the earlier mentioned debate about non-directiveness. In the era of the new genetics, decisions about genetic tests more and more may come to be regarded as medical, rather than as personal decisions. To be sure, the protocol explained above is supposed to apply for all pre-symptomatic tests, but in the case of FAP ‘things are really different’, as the clinical geneticist readily admits:

With FAP the consequences are very straightforward indeed. Thus, we advise people to follow the intended course, but counsellors may often deviate from the protocol. If people have made up their mind and feel no need to talk to the psychologist, we may take blood for example after the first session. It really depends on the circumstances. . . . In keeping track of families however, clinical genetics is more reticent and less directive than the Foundation (for the Detection of Hereditary Tumours). When counselling people, we always make clear that they are
the only ones who can inform their relatives and that it is really important to do that. What we do not however, is checking whether that is really being done. As clinical geneticists, we sometimes perhaps have a bit of a holier-than-thou attitude. (An interview with a clinical geneticist at a regional clinical genetics centre)

Thus what we find in the FAP-network is not only co-existence, but also interference between two different regimes, configuring in different ways the autonomy of patients and individuals at risk. In a regime of prevention, everything is done in order to promote health. In this context, the notion of individual autonomy mainly appears as an opportunity or boundary that one should respect (days are gone in which the doctor knew best). In a regime of self-determination everything is done to uphold freedom of choice, although one need not always have a ‘holier-than-thou’ attitude (sometimes choices may be straightforward indeed).

Conclusion

In the foregoing we have discussed the development of a Dutch FAP-network as an example of emergent practices of ‘new genetics’ in which individuals are not only presented with information about future health risks, but also with related options for prevention. In the context of this new genetics new questions and challenges arise especially in regard to the issue of non-directiveness. We have seen that counselling is generally emphasized as a crucially important tool in dealing with these challenges. The quality of communication and recommendations in conversations between health care providers and their clients is considered all-important for the way clients will cope with options for screening and preventive interventions. From this point of view, it has been argued by Resnik (2003), for example, that the need for advice has much more urgency and importance as the information obtained from genetic tests is becoming more complex and the options for treatment and prevention are increasing. Patients ‘now more than ever ... need advice that they can trust’ (Resnik, 2003, p. 253). The notion of trust we see as especially interesting here, because trust obviously may come from many more sources than information alone. Trust, that is, need not be exclusively grounded in the personal interactions between health care workers and their clients, but may also be embodied in a whole network of actors, technologies, procedures, standards and routines, securing the effectiveness and reliability of preventive screening and interventions (Boenink, 2003; Schermer, 2001).

This is the point we have emphasized in terms of our own co-evolutionary perspective. By taking into account the whole network constituting a medical practice of cancer genetic screening, the course of action of an individual at risk does not appear as the outcome of one particular instance of autonomous decision-making, but as being shaped by various activities that make up a medical regime. In our discussion of the emerging FAP-network, we have focused in particular on the various and conflicting ways in which the autonomy of patients and individuals at risk is being shaped or configured within two different regimes that we have
described in terms of prevention and self-determination. Interference between these two regimes we see as an important characteristic of the emergent new genetics, implicating indeed a growing tension between directivity and non-directivity. What our analysis suggests, however, is that we should not discuss this tension only as a challenge to be dealt with in terms of a particular ethos of counselling, but should extend our view to the whole complex of elements and activities which shapes individual autonomy in the context of different regimes. How, in other words, interfering regimes and tensions between these different regimes will be made productive or perhaps even counterproductive, will remain an important point of concern in the new genetics of the future.

Acknowledgements

The authors would like to thank Brenda Diergaarde for her contribution to the research that was performed in order to write this article. Also, we would like to thank our colleagues Hans Harbers, Annemarie Mol, Rita Struhkamp and the two anonymous reviewers for their helpful comments on earlier versions of this article.

Notes

1. According to the Code of Ethics of the (American) National Society of Genetic Counsellors (1993), genetic counsellors should ‘enable their patients to make informed, independent decisions, free from coercion, by providing or illuminating the necessary facts and clarifying the alternatives and anticipated consequences’ (quoted by Resnik, 2003, p. 246).
2. This debate about the merits and limits of non-directiveness also concerns another cornerstone of the ethos of genetic counselling, that is, the principle of confidentiality (see Biesecker, 1998; de Wert, 1999; and Resnik, 2003).
3. Our co-evolutionary approach to the issues of autonomy and non-directiveness has also been inspired by a study of Maartje Schermer (2001) in which she discusses the ‘different faces of autonomy’ in the everyday practices of hospital care.
4. For a historical reconstruction of the practices which constituted FAP as a hereditary disease, see Palladino (2002).
5. One year later a similar proposal was published by a number of Dutch physicians in the American Journal of Medicine, focusing on another hereditary cancer syndrome (MEN type 2A). The authors observed that ‘at present, coordination of the administrative handling of these large groups of patients and their relatives does not exist in any country’. In their view, preventive medicine involving large patient groups needed a collective effort, because ‘this responsibility cannot be borne by individual physicians’ (Lips et al., 1982, pp. 305–6).
6. In practice, however, the option of prenatal diagnosis appears to be rarely used (Whitelaw et al., 1996).
7. For a recent discussion of the meaning and use of ‘regime’ concepts in the study of socio-technical developments, see Deuten (2003). He defines the notion of regime as referring to ‘complexes of (more or less) shared rules on how to act and interact, and to a system of corresponding interdependencies between (groups of) actors’ (Deuten, 2003, p. 31). He also refers to the work of van de Poel (1998, p. 13) who notes that ‘what is crucial for the genesis of technological regimes is that actors at the local level interact and react to each other, creating interdependencies and so emergently a global level of artefacts, design tools,
technical norms and the like, which then enable and constrain further action at the local level’.

For another elaborate discussion, see Rip and Kemp (1998).

8. This does not imply that other medical practices are naturally directive. What we argue here is the particular emphasis on patient autonomy in the definition and practice of clinical genetics (see also Note 1). Whether a non-directive approach is actually possible is another matter. For critical reviews of non-directiveness and neutrality, see van Zuuren (1996, 1997), Steendam (1996), and Michie et al. (1997).

9. The history of this clinical genetic regime is strongly related with the advent of prenatal diagnosis and the option of selective abortion, which made autonomous and informed decision-making by individual parents a particular sensitive issue. In the Netherlands, this clinical genetic regime served as a ‘niche’ in which, in the 1980s, the first new options for DNA-diagnosis further shaped the practice of clinical genetics as a regime of self-determination, insofar as these options primarily brought along new opportunities for (reproductive) choice. For an elaborate historical account of how, in the Netherlands, clinical genetics emerged as a ‘regime’, see Nelis (1998, 1999).

10. For other studies focusing on the multiple manifestations of autonomy in daily care practices, see Pols (2003) and Struhkamp (2004).

11. The ‘configuration’ of autonomy we may see as a particular aspect of what Steve Woolgar has denoted as the configuration of (putative) users in practices of design, by defining their identities and setting constraints upon their likely future actions. See Woolgar (1991). For similar analyses of the various ways in which subjects are framed or defined by medical practices, see Dodier (1998) and Parthasarathy (2005).

12. The protocol which informs the practice of clinical genetics was developed in response to the first option for DNA diagnosis that was introduced at the end of the 1980s for Huntington’s disease (Nelis, 1998).

References


Dudok de Wit, A.C. (1997) To Know or Not to Know: The Psychological Implications of Presymptomatic DNA-Testing for Autosomal Dominant Late Onset Disorders (Leiden: University of Leiden).


